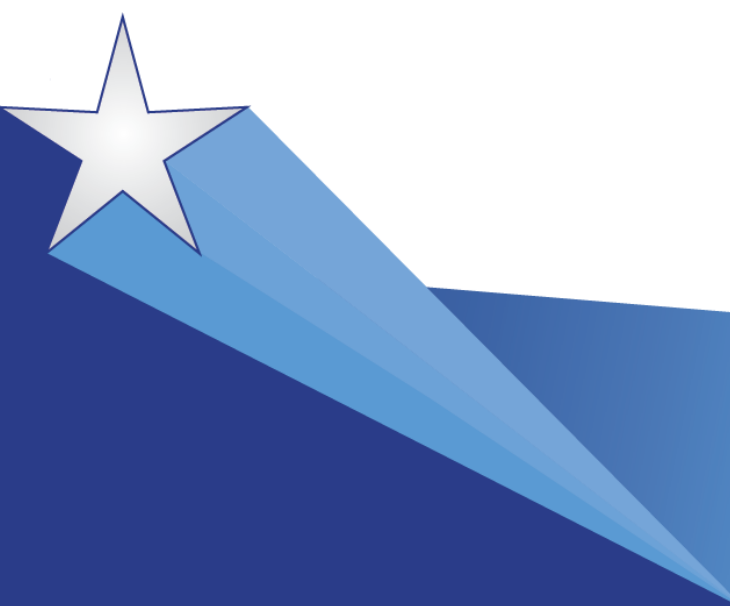




S of DNA:

Unraveling the Mysteries of Genetics Information for Consumers

A decorative graphic in the bottom left corner consisting of a white five-pointed star with a blue outline, set against a blue background that tapers into a white background.

Carolyn Martin, MLS, AHIP
Consumer Health Coordinator
National Network of Libraries of Medicine
Pacific Northwest Region (NN/LM PNR)
martinc4@uw.edu

Agenda

- Basic genetics overview
- Genetic health literacy & genetic science literacy
- Genetic testing including direct-to-consumer
- Genetic consumer health resources
- Ethics and privacy
- Precision Medicine Initiative



[Presentation resources](https://nnlm.gov/pnr/guides/training-resources-you-can-use/presentations)

<https://nnlm.gov/pnr/guides/training-resources-you-can-use/presentations>

Who We Are



NIH

NLM

NN/LM

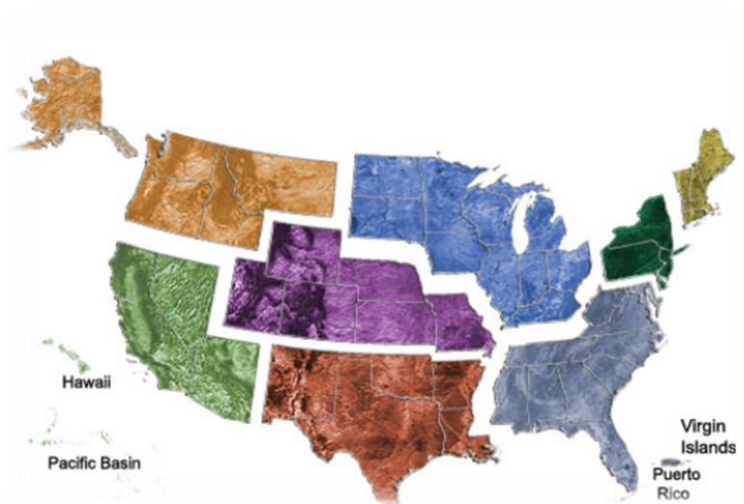
What's the difference?





The mission of NN/LM is to advance the progress of medicine and improve the public health by:

- Providing all U.S. health professionals with equal access to biomedical information.
- Improving the public's access to information to enable them to make informed decisions about their health.



[NN/LM PNR](https://nnlm.gov/pnr)

<https://nnlm.gov/pnr>

News Headlines

Scientists Say They Hope To Create A
Human Genome In The Lab

Humans will be 'irrevocably altered' by genetic editing, warn scientists ahead of summit

An open letter from 150 scientists, campaigners and health experts is calling for a worldwide ban on genetic editing ahead of a summit in Washington

**The Genetic Tool That Will
Modify Humanity**

Crispr allows scientists to control the blueprints of life, for better or worse.

British Scientists Seek Permission To Edit
DNA In Human Embryos

Opioids: Can a Genetic Test
Identify an Addict in the
Making?

Genetically Modified Humans?
How Genome Editing Works

Birth of Baby With Three Parents' DNA Marks Success for Banned Technique

Consumer Genomic Health Literacy

- Lack biology basics
- Lack mathematical concepts
- Low health literacy



Definitions

- Genomic Health Literacy
 - The capacity to obtain, process, understand, and use genomic information for health related decision making.
- Genomic Science Literacy
 - The knowledge of basic genetics and genomics concepts and processes needed to build conceptual understanding, and the necessary mathematical knowledge to support this comprehension.



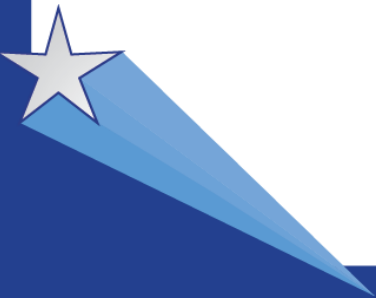
Leading causes of death

1. Heart disease: 614,348
2. Cancer: 591,699
3. Chronic lower respiratory diseases: 147,101
4. Accidents (unintentional injuries): 136,053
5. Stroke (cerebrovascular diseases): 133,103
6. Alzheimer's disease: 93,541
7. Diabetes: 76,488
8. Influenza and pneumonia: 55,227
9. Nephritis, nephrotic syndrome, and nephrosis: 48,146
10. Intentional self-harm (suicide): 42,773

The Story of You

[The Story of You](https://www.youtube.com/watch?v=TwXXgEz9o4w)

<https://www.youtube.com/watch?v=TwXXgEz9o4w>



CATEGORIES OF DISEASES ATTRIBUTED TO GENES

- Chromosomal Diseases
- Monogenic Diseases
- Multifactorial Diseases



Genetic Testing

including Direct-to-Consumer



Genetic Testing








- Analysis using small samples of blood or body tissue
- Determines whether individual carries genes for certain inherited disorders
- Can reveal presence, absence, or malformation of genes or chromosomes
- Complex tests don't yield easy-to-understand results

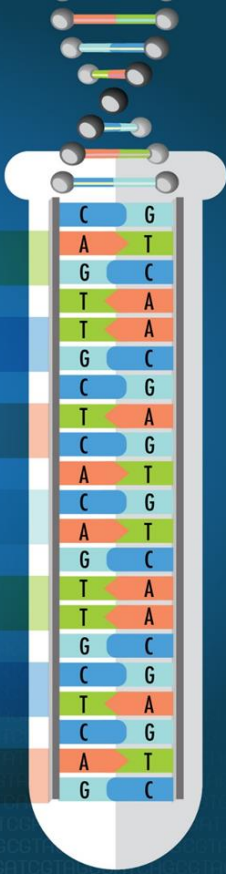


Clinical Uses of Genetic Tests

GENETIC TESTING
NHGRI FACT SHEETS
genome.gov

Genetic Tests Can Help to:

-  **Diagnose Your Disease**
-  **Pinpoint Genetic Factors That Caused Your Disease**
-  **Predict How Severe Your Disease Might Be**
-  **Choose the Best Medicine and Correct Dose**
-  **Discover Genetic Factors That Increase Your Disease Risk**
-  **Find Genetic Factors That Could Be Passed to Your Children**
-  **Screen Newborns for Certain Treatable Conditions**



NIH National Human Genome Research Institute

[Genetic Testing image from Genome.gov](https://www.genome.gov/images/content/genetic_testing.jpg)

https://www.genome.gov/images/content/genetic_testing.jpg

Jean's Genetic Testing Timeline

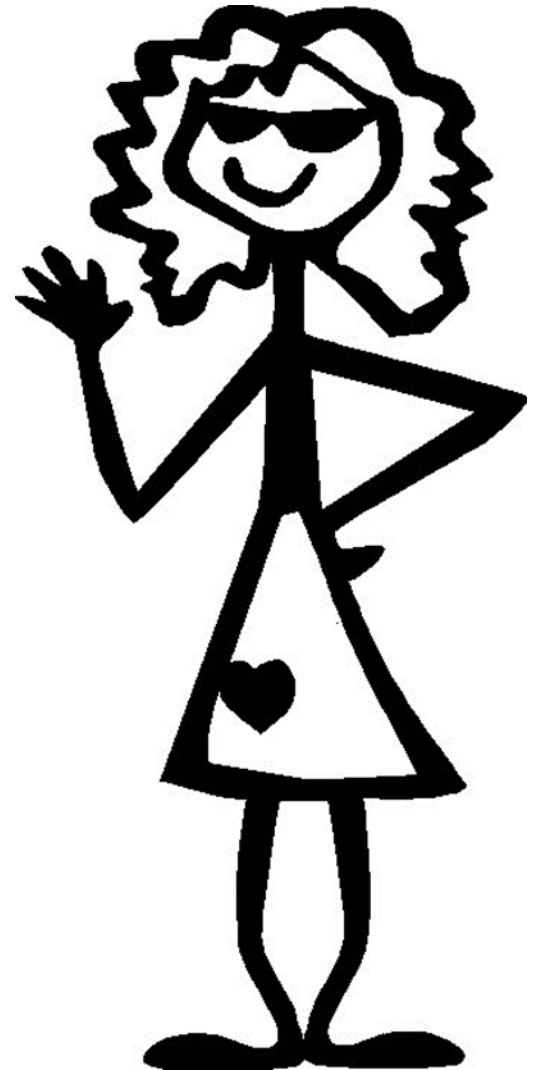
Age 1 Day: newborn testing for a few serious childhood diseases

Age 30: carrier testing (with her partner) before getting pregnant

Age 35: predictive testing when sister develops breast cancer at a young age

Age 45: direct-to-consumer testing to investigate ancestry

Age 65: pharmacogenomics testing when Plavix (anti-platelet drug) was not effective



Genetic Testing Results

What genes and what variants did you test for?

- Different tests offered for the same conditions.
- Knowledge always changing.

Might not have enough examples in the database to determine associations between specific variants and specific conditions.

Might not have enough examples of people like you in the database.

Possibility of false positive and false negative results.



BRCA1

BRCA2



- Majority of breast and ovarian cancers are not linked to BRCA
- Only 0.2% carry BRCA mutations
- U.S. Preventive Services Task Force recommends that women who have family members with breast, ovarian, tubal, or peritoneal cancer be assessed
- Women who are found to have a family history that may be associated with BRCA1 or BRCA2 mutations should receive genetic counseling and subsequent BRCA testing, if indicated
- Having the mutation does not necessarily mean cancer will develop, but it does increase risk

[Lab Tests Online BRCA information](https://labtestsonline.org/news/150409brca/)

<https://labtestsonline.org/news/150409brca/>

[NCI BRCA Fact Sheet](https://www.cancer.gov/about-cancer/causes-prevention/genetics/brca-fact-sheet#q1)

<https://www.cancer.gov/about-cancer/causes-prevention/genetics/brca-fact-sheet#q1>

Genetic Testing- is it necessary?



The screenshot shows the 'Consumer Reports Health Choices' website. The main navigation bar includes links for Home, About, Partnerships, Campaigns, Catalog, In Depth, Connect, News and notes, and Patients' stories. The 'Catalog item' section displays the title 'Making Smart Decisions About Genetic Testing (ACMG)'. Below the title, it lists details: Campaign Series: Choosing Wisely, Medical Category: Tests, Article Type: Advice, Language: Plain English, Spanish, Affiliation: American College of Medical Genetics and Genomics, Format: HTML, PDF, and Most recent update: 10/07/2015. A small thumbnail image of the article is shown. To the right, there is a 'Customize the catalog' section with filters for Campaign series, Affiliation, Medical category, Article type, Available Format, and Available Language. At the bottom, there is a 'Files to download' section with links for Plain English and Spanish versions of the article, and a 'Webpage file' link.

ConsumerReportsHealth

Search

ConsumerHealthChoices

Home About Partnerships Campaigns Catalog In Depth Connect News and notes Patients' stories

Catalog item

Making Smart Decisions About Genetic Testing (ACMG)

Campaign Series: Choosing Wisely
Medical Category: Tests
Article Type: Advice
Language: Plain English, Spanish
Affiliation: American College of Medical Genetics and Genomics
Format: HTML, PDF
Most recent update: 10/07/2015

Customize the catalog

Filter by Campaign series
Any Campaign series...

Filter by Affiliation
Any Affiliation...

Filter by Medical category
Any Medical category...

Filter by Article type
Any Article type...

Filter by Available Format
Any Available Format...

Filter by Available Language
Any Available Language...

Submit Reset

Files to download

Plain English: [Making Smart Decisions About Genetic Testing \(ACMG\) -->](#)
Spanish: [Cómo tomar decisiones inteligentes sobre las pruebas genéticas -->](#)
See webpage version

Webpage file: [Making Smart Decisions About Genetic Testing -->](#)
See webpage version

Webpage file: [Cómo tomar decisiones inteligentes sobre las pruebas genéticas -->](#)

Questions to ask:

- Am I in the group at risk and should I get tested?
- If I decide to get tested, what do the results mean?
- What are my treatment options based on results?
- How do I decide on treatment?

[Choosing Wisely](#)

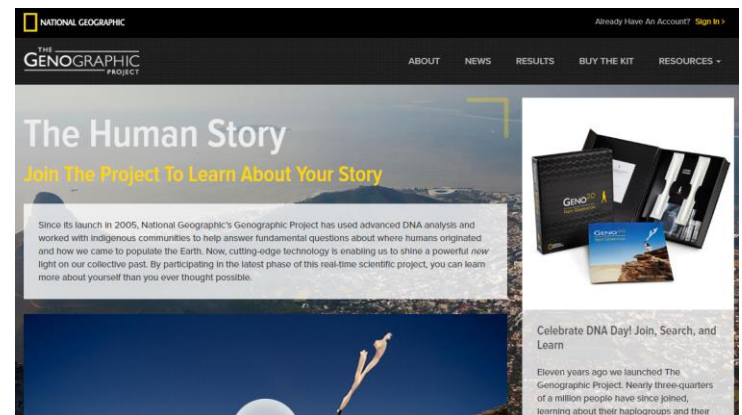
<http://consumerhealthchoices.org/catalog/making-smart-decisions-about-genetic-testing-acmg/>

Genetic Counseling/Consultation

- Evaluating family history and medical records
- Ordering genetic tests
- Identify and interpret risks of inherited disorders, explain inheritance patterns
- Helping people understand and reach decisions about what to do next




Direct to Consumer Testing




Genomic Testing- Athletic Ability

- Over 36 companies marketing genetic tests
- Endurance and power
- Poor quality control
- Targeted to coaches and parents
- Individuals also wanting to focus training


Achieve
your full potential.



Increase
your athletic performance.



Harness
your natural ability through
personalized genomics.


www.GenomicExpress.com

Genomic Testing- Consensus Statement

Consensus statement



OPEN ACCESS

Direct-to-consumer genetic testing for predicting sports performance and talent identification: Consensus statement

Nick Webborn,¹ Alun Williams,² Mike McNamee,³ Claude Bouchard,⁴ Yannis Pitsiladis,⁵ Ildus Ahmetov,⁶ Euan Ashley,⁷ Nuala Byrne,⁸ Silvia Camporesi,⁹ Malcolm Collins,¹⁰ Paul Dijkstra,¹¹ Nir Eynon,¹² Noriyuki Fuku,¹³ Fleur C Garton,¹⁴ Nils Hoppe,¹⁵ Søren Holm,¹⁶ Jane Kaye,¹⁷ Vassilis Klissouras,¹⁸ Alejandro Lucia,¹⁹ Kamiel Maase,²⁰ Colin Moran,²¹ Kathryn N North,¹⁴ Fabio Pigozzi,²² Guan Wang⁵

► Additional material is published online only. To view please visit the journal online (<http://dx.doi.org/10.1136/bjsports-2015-095343>).

For numbered affiliations see end of article.

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Accepted 25 September 2015

ABSTRACT

The general consensus among sport and exercise genetics researchers is that genetic tests have no role to play in talent identification or the individualised prescription of training to maximise performance. Despite the lack of evidence, recent years have witnessed the rise of an emerging market of direct-to-consumer marketing (DTC) tests that claim to be able to identify children's athletic talents. Targeted consumers include mainly coaches and parents. There is concern among the scientific community that the current level of knowledge is being misrepresented for commercial purposes. There remains a lack of universally accepted guidelines and legislation for DTC testing in relation to all forms of genetic testing and not just for talent identification. There is concern over the lack of clarity of information over which specific genes or variants are being tested and the almost universal lack of appropriate genetic counselling for the interpretation of the genetic data to consumers. Furthermore independent studies have identified issues relating to quality control by DTC laboratories with different results being reported from

of the evidence in relation to genetic testing and the limitations of current knowledge. This article reviews the issues around the currently available evidence behind the genetic testing, comments on the ethical considerations and makes recommendations about such tests.

STATEMENT ON BACKGROUND TO THE CONSENSUS PROCESS

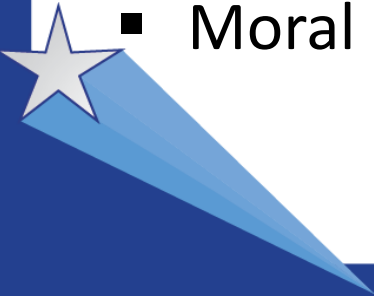
A group of world experts in the field of genomics, exercise, sport performance, disease, injury and antidoping gathered with the International Federation of Sports Medicine (FIMS) Scientific Commission for a symposium to discuss the current state of knowledge and to share ideas. One key concern was the misuse of research evidence and the misinformation about genetic testing, particularly when marketed directly to the public, coaches or parents. This is known as DTC testing for the purpose of talent identification and to assess potential for future sports performance. There have been

Concerns

- Privacy
- Legality
- Who has access?
- How useful now?
- What all is being done now and in the future with the information?
- Unexpected surprises?
- Test results can vary among companies
- Validity of tests
- No counseling provided
- Who can get the testing?

Benefits

- Learn more about own health
- More effective medical treatments
- Learn more about ethnicity and family history
- Bring awareness to family health issues for future generations
- Motivation to work on health habits
- Encourages patient engagement
- Contributing to advancement of healthcare and science
- Moral obligation



Genetics Home Reference

NIH U.S. NATIONAL LIBRARY OF MEDICINE



Your Guide to Understanding
Genetic Conditions

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Health Conditions

Genes

Chromosomes & mtDNA

Resources

Help Me Understand Genetics

Home

Help Me Understand Genetics

Genetic Testing


What is direct-to-consumer genetic testing?

What is direct-to-consumer genetic testing?

Traditionally, genetic tests have been available only through healthcare providers such as physicians, nurse practitioners, and genetic counselors. Healthcare providers order the appropriate test from a laboratory, collect and send the samples, and interpret the test results. Direct-to-consumer genetic testing refers to genetic tests that are marketed directly to consumers via television, print advertisements, or the Internet. This form of testing, which is also known as at-home genetic testing, provides access to a person's genetic information without necessarily involving a doctor or insurance company in the process.

If a consumer chooses to purchase a genetic test directly, the test kit is mailed to the consumer instead of being ordered through a doctor's office. The test typically involves collecting a DNA sample at home, often by swabbing the inside of the cheek, and mailing the sample back to the laboratory. In some cases, the person must visit a health clinic to have blood drawn. Consumers are notified of their results by mail or over the telephone, or the results are posted online. In some cases, a genetic counselor or other healthcare provider is available to explain the results and answer questions. The price for this type of at-home genetic testing ranges from several hundred dollars to more than a thousand dollars.

For more information about direct-to-consumer genetic testing:

The American College of Medical Genetics, which is a national association of doctors specializing in genetics, has issued [a statement on direct-to-consumer genetic testing](#) .

The American Society of Human Genetics, a professional membership organization for specialists in genetics, has also issued [a statement on direct-to-consumer genetic testing in the United](#)

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Genetic Testing

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Research <ul style="list-style-type: none">Statistics and ResearchClinical TrialsJournal Articles	Resources <ul style="list-style-type: none">Reference DeskFind an Expert	For You <ul style="list-style-type: none">Patient Handouts

Summary

Genetic tests are tests on blood and other tissue to find **genetic disorders**. Over 2000 tests are available. Doctors use genetic tests for several reasons. These include

- Finding genetic diseases in unborn babies
- Finding out if people carry a gene for a disease and might pass it on to their children
- Screening embryos for disease
- Testing for genetic diseases in adults before they cause symptoms
- Making a diagnosis in a person who has disease symptoms
- Figuring out the type or dose of a medicine that is best for a certain person

People have many different reasons for being tested or not being tested. For some, it is important to know whether a disease can be prevented or treated if a test is positive. In some cases, there is no treatment. But test results might help a person make life decisions, such as family planning or insurance coverage. A **genetic counselor** can provide information about the pros and cons of testing.

NLM: National Human Genome Research Institute

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MEDICAL ENCYCLOPEDIA

BRCA1 and BRCA2 gene testing
Buccal smear
Genetic testing and your cancer risk

Related Health Topics

Birth Defects

FEDERAL TRADE COMMISSION ESPAÑOL

CONSUMER INFORMATION

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Direct-to-Consumer Genetic Tests

Could a simple medical test tell you if you are likely to get a particular disease? Could it evaluate your health risks and even suggest a specific treatment? Could you take this test in the privacy of your home, without a doctor's prescription or guidance?

Some companies say genetic testing can do all this and more. They claim that direct-to-consumer (DTC) genetic testing can screen for diseases and provide a basis for choosing a particular diet, dietary supplement, lifestyle change, or medication. These companies primarily sell their tests online and through multi-level marketing networks.

The Federal Trade Commission (FTC) wants you to know the facts about the DTC marketing of genetic tests.

Related Items

[Anatomy of a Cancer Treatment Scam](#)

- Dietary Supplements
- Miracle Health Claims
- Cancer Treatment Scams

[Genes and Genetic Tests](#)
[Interpreting the Results](#)
[Company Claims](#)
[If You're Considering a DTC Genetic Test](#)
[For More Information](#)

American College of Medical Genetics and Genomics

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ACMG STATEMENT | Genetics
inMedicine

Direct-to-consumer genetic testing: a revised position statement of the American College of Medical Genetics and Genomics

ACMG Board of Directors¹

Disclaimer: These recommendations are designed primarily as an educational resource for medical geneticists and other health-care providers to help them provide quality medical genetics services. Adherence to these recommendations does not necessarily assure a successful medical outcome. These recommendations should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonably directed to obtaining the same results. In determining the propriety of any specific procedure or test, geneticists and other

clinicians should apply their own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. It may be prudent, however, to document in the patient's record the rationale for any significant deviation from the recommendations.

Genet Med advance online publication 17 December 2015

Key Words: consumer; direct-to-consumer; genetic testing; self-testing; OTC

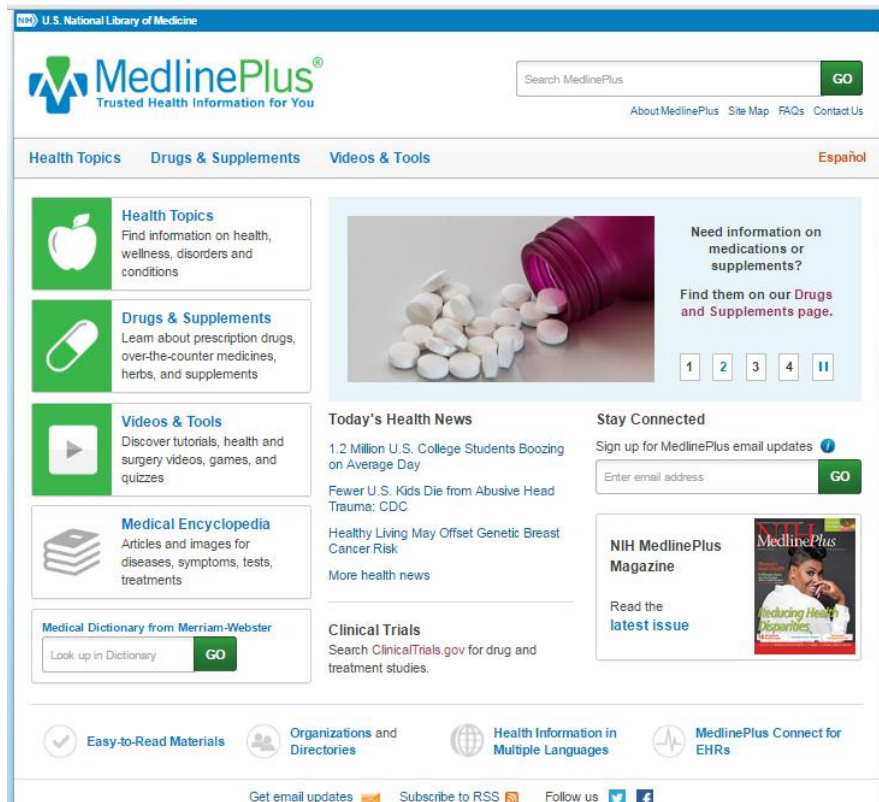
With ongoing genetic discoveries and improvements in technology, more genetic tests are available than ever before. Along with greater availability has come increased consumer demand for genetic tests and expansion of direct-to-consumer testing. The American College of Medical Genetics and Genomics (ACMG) has revised its 2008 e-publication regarding this issue (ACMG Statement on Direct-to-Consumer Genetic Testing, retired; available by request to acmg@acmg.net) and believes that it is critical for the public to realize that genetic testing is only one part of a complex process that includes genetic risk

- A genetics expert such as a certified medical geneticist or genetic counselor should be available to help the consumer determine, for example, whether a genetic test should be performed and how to interpret test results in light of personal and family history. A board-certified genetic counselor can help facilitate this process by providing information about the test and helping to explain test results. A number of risks can be reduced if a board-certified genetics professional is involved in genetic testing, including inadequate or lack of informed consent.

Consumer Resources



MedlinePlus



- Section: Genetics/Birth Defects
- Health Topic pages:
 - Genetics
 - Genetic testing
 - Genetic counseling
 - Genetic disorders
 - Genes and gene therapy
- text word search

[MedlinePlus](https://medlineplus.gov/) <https://medlineplus.gov/>

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Genetics/Birth Defects

[Abnormalities](#) *see* [Birth Defects](#)

[Achondroplasia](#) *see* [Dwarfism](#)

[Adrenoleukodystrophy](#) *see* [Leukodystrophies](#)

[Alpha-1 Antitrypsin Deficiency](#)

[Amniocentesis](#) *see* [Prenatal Testing](#)

[Anencephaly](#) *see* [Neural Tube Defects](#)

[Arnold-Chiari Malformation](#) *see* [Chiari Malformation](#)

[Ataxia](#) *see* [Friedreich's Ataxia](#)

[Ataxia Telangiectasia](#)

[Birth Defects](#)

[Blood Coagulation Disorders](#) *see* [Hemophilia](#)

[Brain Disorders, Inborn Genetic](#) *see* [Genetic Brain Disorders](#)

[Brain Malformations](#)

[Canavan Disease](#) *see* [Leukodystrophies](#)

[Cephalic Disorders](#) *see* [Brain Malformations](#)

[Cerebral Palsy](#)

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Genetic Disorders

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<ul style="list-style-type: none"> Summary Start Here Diagnosis and Tests Treatments and Therapies 	<ul style="list-style-type: none"> Living With Related Issues Specifics Genetics 	<ul style="list-style-type: none"> No links available

Research	Resources	For You
<ul style="list-style-type: none"> Statistics and Research Clinical Trials Journal Articles 	<ul style="list-style-type: none"> Reference Desk Find an Expert 	<ul style="list-style-type: none"> Children Teenagers Women Patient Handouts

Summary

Genes are the building blocks of heredity. They are passed from parent to child. They hold DNA, the instructions for making proteins. Proteins do most of the work in cells. They move molecules from one place to another, build structures, break down toxins, and do many other maintenance jobs.

Sometimes there is a mutation, a change in a gene or genes. The mutation changes the gene's instructions for making a protein, so the protein does not work properly or is missing entirely. This can cause a medical condition called a genetic disorder.

You can inherit a gene mutation from one or both parents. A mutation can also happen during your lifetime.

There are three types of genetic disorders:

Single gene disorder, where a single gene is affected. Sickle cell anemia is an example.

MEDICAL ENCYCLOPEDIA

[Alström syndrome](#)

[Basal cell nevus syndrome](#)

[Beriberi](#)

[Cystinuria](#)

[Genetics](#)

[Hemochromatosis](#)

Diagnosis and Tests

- How Are Genetic Conditions Diagnosed? [\(NIH\)](#) (National Library of Medicine)
- MedlinePlus: Newborn Screening [\(NIH\)](#) (National Library of Medicine)
Available in Spanish
- Progeria Research Foundation Diagnostic Testing Program (Progeria Research Foundation, Inc.)

Treatments and Therapies

- How Are Genetic Conditions Treated or Managed? [\(NIH\)](#) (National Library of Medicine)

Living With

- Physical Therapy and Occupational Therapy in Progeria (Progeria Research Foundation, Inc.) - PDF

Related Issues

- Learning about an Undiagnosed Condition in an Adult [\(NIH\)](#) (National Human Genome Research Institute)

Specifics

- Bloom's Syndrome (Chicago Center for Jewish Genetic Disorders)
- Chromosomal Conditions (March of Dimes Birth Defects Foundation)
Available in Spanish
- Cryopyrin-Associated Autoinflammatory Syndromes (CAPS) - Juvenile (American College of Rheumatology)
- Genetics Home Reference [\(NIH\)](#) (National Library of Medicine)
- Learning about Poland Anomaly [\(NIH\)](#) (National Human Genome Research Institute)
- Learning about Progeria [\(NIH\)](#) (National Human Genome Research Institute)
- Noonan Syndrome (Mayo Foundation for Medical Education and Research)
- Progeria 101/FAQ (Progeria Research Foundation, Inc.)
- Specific Genetic Disorders [\(NIH\)](#) (National Human Genome Research Institute)
- Triple X Syndrome (Mayo Foundation for Medical Education and Research)
- Williams Syndrome [\(NIH\)](#) (National Institute of Neurological Disorders and Stroke)

Genetics

- Genetics Home Reference: Genetic Conditions [\(NIH\)](#) (National Library of Medicine)

Statistics and Research

Cystic Fibrosis

Down Syndrome

Dwarfism

Fragile X Syndrome

Genetic Brain Disorders

Genetic Counseling

Genetic Testing

Hemochromatosis

Leukodystrophies

Osteogenesis Imperfecta

Prader-Willi Syndrome

Rare Diseases

Sickle Cell Anemia

National Institutes of Health

The primary NIH organization for research on Genetic Disorders is the National Institute of Child Health and Human Development

NIH MedlinePlus Magazine

Medical Mysteries: "Thankful They Found a Cause"

Medical Mysteries: "We Feel Deep Compassion for Patients..."

Medical Mysteries: NIH Clinical Center: There's No Other Hospital Like It

Medical Mysteries: NIH Undiagnosed Diseases Program

Promise and Payoff of Rare Diseases Research

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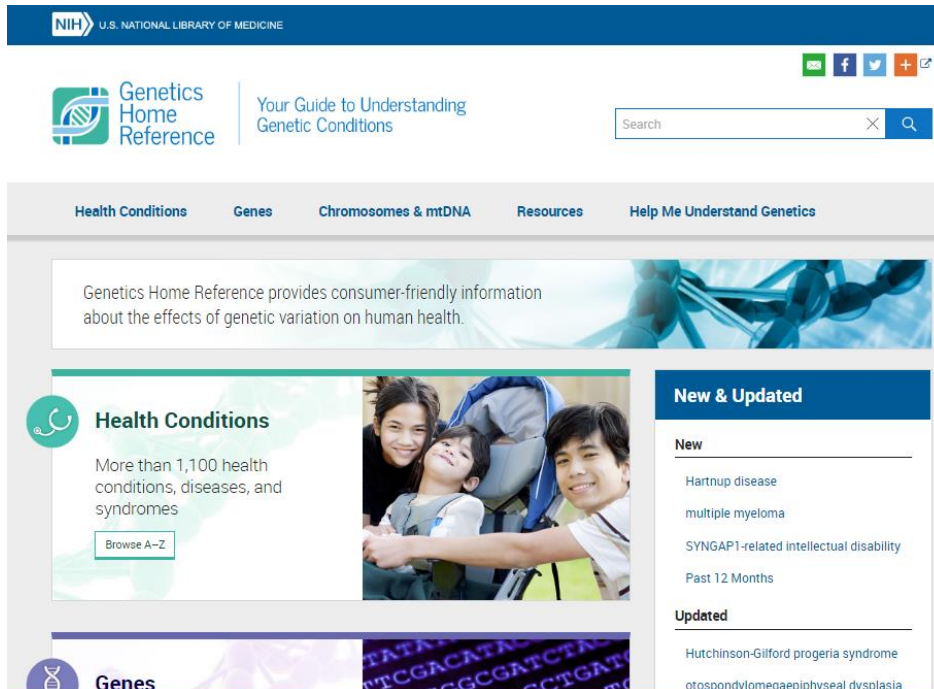
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- ☐ [External Health Links](#) (873)
- ☐ [Drugs and Supplements](#) (10)
- ☐ [Medical Encyclopedia](#) (11)
- ☐ [Videos and Tutorials](#)
- ☐ [News](#) (4)
- ☐ [MedlinePlus Magazine](#) (23)
- ☐ [Other Resources](#) (21)
- ☐ [Multiple Languages](#)

Results 1 - 10 of 945 for **genomics**

1. **Frequently Asked Questions about Genetic and Genomic Science**  (National Human Genome Research Institute)
... this page Frequently Asked Questions About Genetic and **Genomic** Science What are genetics and **genomics**? Why are ... **genomic** technologies? Additional Resources What are genetics and **genomics**? Genetics is a term that refers to the ... <https://www.genome.gov/19016904> - External Health Links
2. **Genomics and Health Impact Update** (Centers for Disease Control and Prevention)
... Knowledge Base. What's New in the Public Health **Genomics** Knowledge Base New Implementation Tools Alzheimer's Disease Cardiomyopathy HIV/AIDS About the **Genomics** & Health Impact Update The Office of Public Health ... www.cdc.gov/genomics/update/current.htm - External Health Links
3. **Brief Guide to Genomics: DNA, Genes and Genomes**  (National Human Genome Research Institute)
... Breve guía de genómica A Brief Guide to **Genomics** DNA, Genes and Genomes Deoxyribonucleic acid (DNA) is ... genetic basis for health and disease. Implications of **Genomics** for Medical Science Virtually every human ailment has ... <https://www.genome.gov/18016863> - External Health Links
4. **Genomic Testing** (Centers for Disease Control and Prevention)


Genetics Home Reference




- Health conditions
- Genes
- Chromosomes and DNA
- Resources
- Genetic handbook

[Genetics Home Reference](https://ghr.nlm.nih.gov/) <https://ghr.nlm.nih.gov/>

Genetics Home Reference



Health ConditionsGenesChromosomes & mtDNAResourcesHelp Me Understand Genetics



Health Conditions

Explore the signs and symptoms, frequency, genetic cause, and inheritance pattern of various conditions, diseases, and syndromes.

Browse by Category

Browse by First Letter

0-9

A

B

C

D

E

F

G

H

I

J

K

L

M

N

O

P

Q

R

S

T

U

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W

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Z

A-[alpha1ipoprotein Neuropathy](#), see [Tangier disease](#)
A-T, see [ataxia-telangiectasia](#)
AAA, see [triple A syndrome](#)
AAA syndrome, see [triple A syndrome](#)
AADC deficiency, see [aromatic L-amino acid decarboxylase deficiency](#)
Aarskog syndrome, see [Aarskog-Scott syndrome](#)
[Aarskog-Scott syndrome](#)
AAS, see [Aarskog-Scott syndrome](#)
AASA dehydrogenase deficiency, see [pyridoxine-dependent epilepsy](#)
Aase syndrome, see [Diamond-Blackfan anemia](#)
Aase-Smith syndrome II, see [Diamond-Blackfan anemia](#)
AAT, see [alpha-1 antitrypsin deficiency](#)

Learn More about Health Conditions

What does it mean if a disorder seems to run in my family?


What are the different ways in which a genetic condition can be inherited?

What are complex or multifactorial disorders?

What does it mean to have a genetic predisposition to a disease?




Genetics Home Reference

Health ConditionsGenesChromosomes & mtDNAResourcesHelp Me Understand Genetics

breast cancer

Print AllOpen AllClose All

- Description
- Frequency
- Genetic Changes
- Inheritance Pattern
- Diagnosis & Management
- Other Names for This Condition
- Additional Information & Resources
- Sources for This Page
- Images

Health ConditionsGenesChromosomes & mtDNAResourcesHelp Me Understand Genetics

breast cancer

Print AllOpen AllClose All

- Description
- Frequency
- Genetic Changes
- ▼ Inheritance Pattern

Most cases of breast cancer are not caused by inherited genetic factors. These cancers are associated with somatic mutations in breast cells that are acquired during a person's lifetime, and they do not cluster in families.

In hereditary breast cancer, the way that cancer risk is inherited depends on the gene involved. For example, mutations in the [BRCA1](#) and [BRCA2](#) genes are inherited in an [autosomal dominant pattern](#), which means one copy of the altered gene in each cell is sufficient to increase a person's chance of developing cancer. Although breast cancer is more common in women than in men, the mutated gene can be inherited from either the mother or the father.

In the other syndromes discussed above, the gene mutations that increase cancer risk also have an autosomal dominant pattern of inheritance. It is important to note that people inherit an increased likelihood of developing cancer, not the disease itself. Not all people who inherit mutations in these genes will ultimately develop cancer.

In many cases of breast cancer that clusters in families, the genetic basis for the disease and the mechanism of inheritance are unclear.

Related Information

- [What does it mean if a disorder seems to run in my family?](#)
- [What are the different ways in which a genetic condition can be inherited?](#)
- More about [Inheriting Genetic Conditions](#)

- Diagnosis & Management

Genetics Home Reference

Health Conditions Genes Chromosomes & mtDNA **Resources** Help Me Understand Genetics

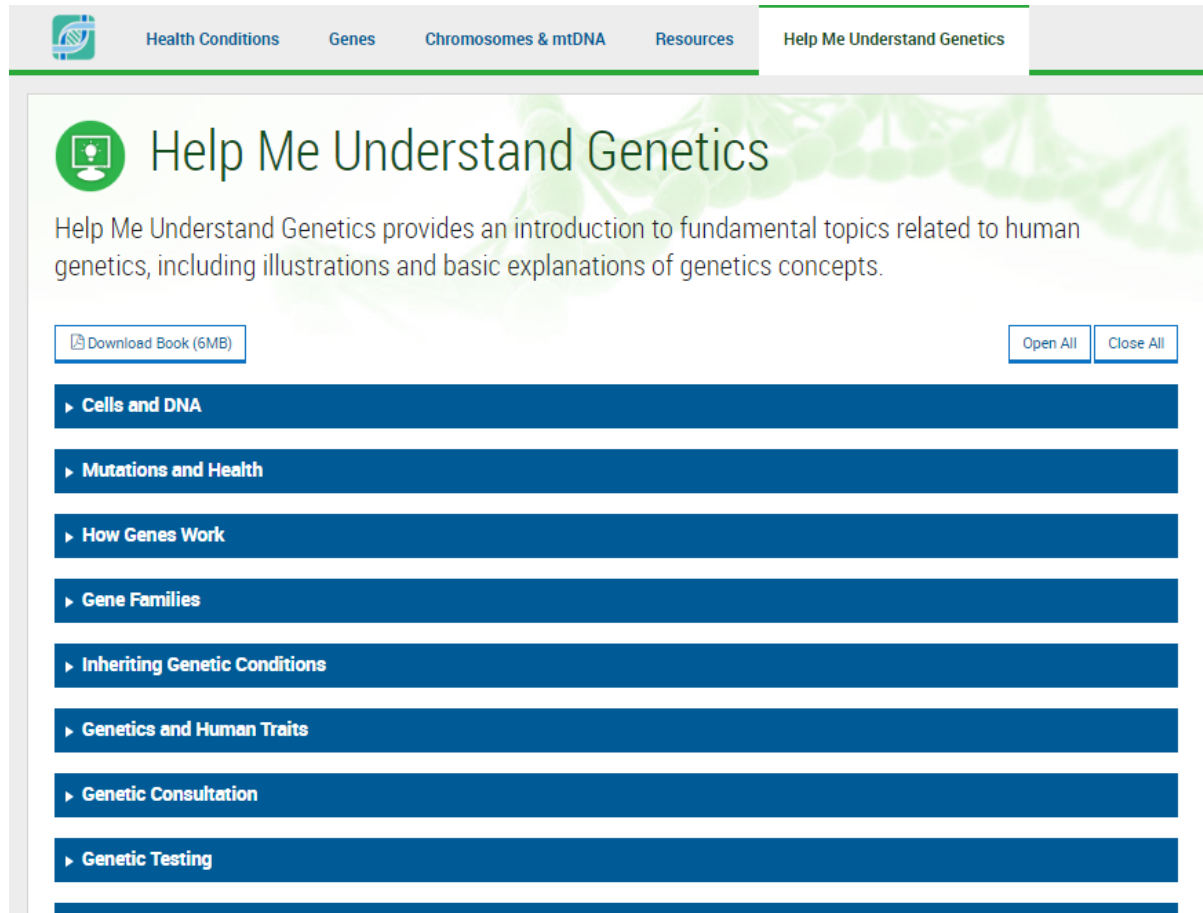
Resources

Each of the following categories provides links to useful genetics resources on the web.

[Open All](#) [Close All](#)

- ▶ Support and Advocacy
- ▶ Financial Assistance
- ▶ General Genetics
- ▶ Genetic Testing
- ▶ Classroom Resources
- ▶ Clinical/Professional Resources
- ▶ Genetics Research
- ▶ Bioinformatics Databases
- ▶ Genetics News


Genetics Home Reference



The screenshot displays the 'Help Me Understand Genetics' page. At the top, a navigation bar includes links for 'Health Conditions', 'Genes', 'Chromosomes & mtDNA', 'Resources', and 'Help Me Understand Genetics'. The main heading is 'Help Me Understand Genetics', accompanied by a lightbulb icon. Below the heading, a descriptive paragraph states: 'Help Me Understand Genetics provides an introduction to fundamental topics related to human genetics, including illustrations and basic explanations of genetics concepts.' A 'Download Book (6MB)' button is located on the left, and 'Open All' and 'Close All' buttons are on the right. A list of topics is presented as blue bars with white text and a right-pointing arrow:

- ▶ Cells and DNA
- ▶ Mutations and Health
- ▶ How Genes Work
- ▶ Gene Families
- ▶ Inheriting Genetic Conditions
- ▶ Genetics and Human Traits
- ▶ Genetic Consultation
- ▶ Genetic Testing





NIH National Human Genome Research Institute




National Human Genome
Research Institute

Search Genome.gov

Español






Research FundingResearch at NHGRIHealthEducationIssuesNewsroomCareersAbout

Health

Information about genetics and genomics, rare diseases, patient care and more


For Patients and the Public



Detailed information about genetic disorders, background on genetic and genomic science, pharmacogenomics, family health history tool and online health resources

- › Community Engagement and Community Health
- › Family History
- › Genetics & Genomics Science & Research
- › Genetic & Rare Diseases Information Center
- › Genomic Medicine and Health Care
- › Online Health and Support Resources
- › Specific Genetic Disorders

For Health Professionals




Genetics and genomics information related to patient management, education, NIH and NHGRI research and ethical, legal and social issues

- › Competency & Curricular Resources
- › Genetics 101
- › Genomic Medicine and Health Care
- › Inter-Society Coordinating Committee (ISCC)
- › New Horizons and Research
- › Patient Management
- › Policy and Ethics Issues

Highlights

NIH awards \$55 million to build million-person precision medicine study



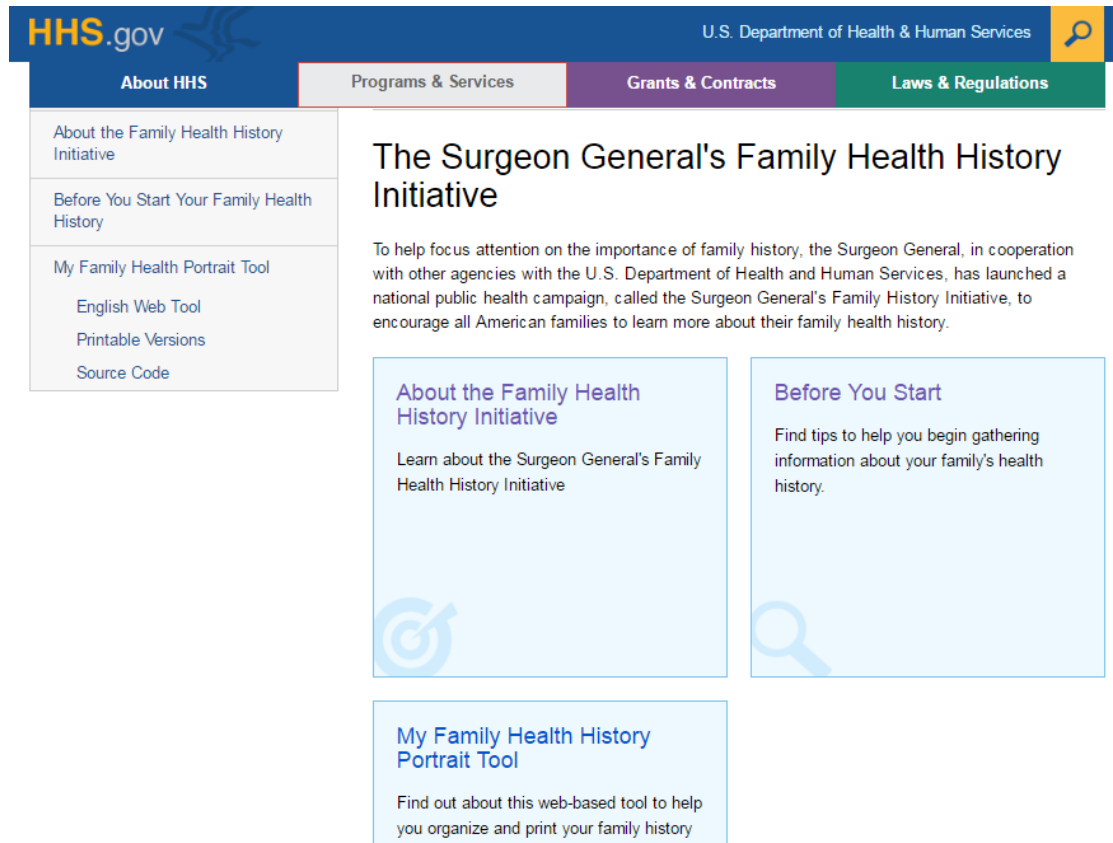
Bethesda, Md., Thurs., July 7, 2016 - The U.S. Food and Drug Administration (FDA) has announced two draft guidances to support President Obama's Precision Medicine Initiative. The guidances will help provide oversight for tests based on next generation sequencing, a technology that examines a person's DNA to detect medically important differences in genomic make-up that could increase the risk for disease.

See Also

- GenomeTV
- Genomic Healthcare Branch
- Fact Sheets
- Genetic Education Resources for Teachers
- All About the Human Genome Project

[NHGRI https://www.genome.gov/](https://www.genome.gov/)

My Family Health Portrait U.S. Surgeon General



The screenshot shows the HHS.gov website with a blue header. The header includes the HHS.gov logo, a search icon, and the text "U.S. Department of Health & Human Services". Below the header is a navigation bar with four tabs: "About HHS" (selected), "Programs & Services", "Grants & Contracts", and "Laws & Regulations".

Under the "About HHS" tab, there is a sidebar with the following links:

- About the Family Health History Initiative
- Before You Start Your Family Health History
- My Family Health Portrait Tool
 - English Web Tool
 - Printable Versions
 - Source Code

The main content area is titled "The Surgeon General's Family Health History Initiative". Below the title is a paragraph:

To help focus attention on the importance of family history, the Surgeon General, in cooperation with other agencies with the U.S. Department of Health and Human Services, has launched a national public health campaign, called the Surgeon General's Family History Initiative, to encourage all American families to learn more about their family health history.

Below the paragraph are three boxes:

- About the Family Health History Initiative**
Learn about the Surgeon General's Family Health History Initiative
- Before You Start**
Find tips to help you begin gathering information about your family's health history.
- My Family Health History Portrait Tool**
Find out about this web-based tool to help you organize and print your family history

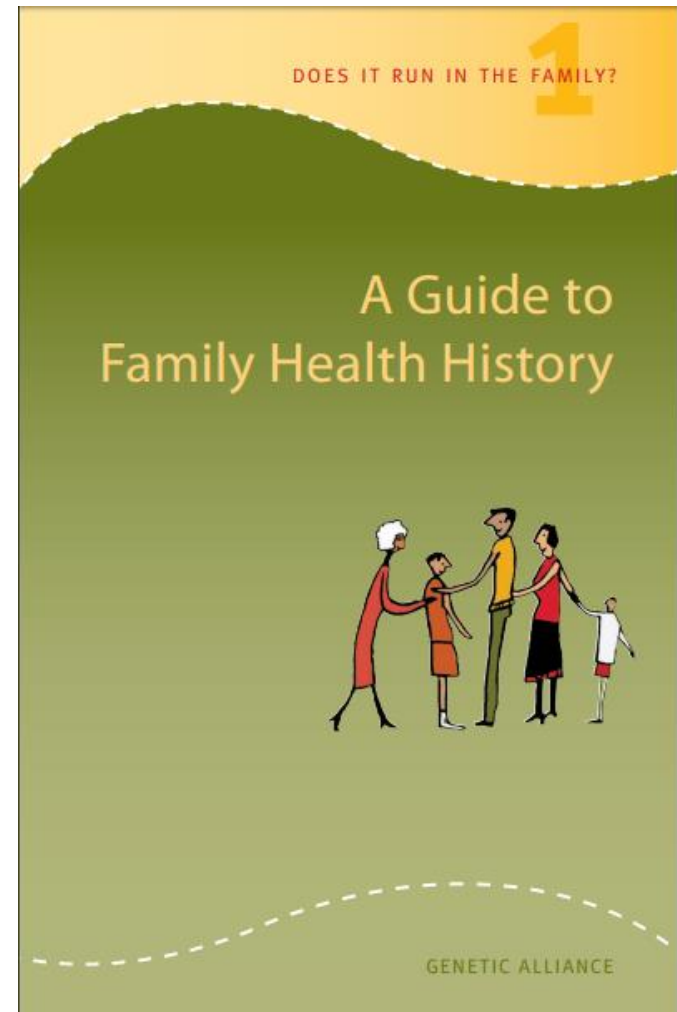
[Surgeon General's Family Health History Initiative:](http://www.hhs.gov/programs/prevention-and-wellness/family-health-history/index.html)

<http://www.hhs.gov/programs/prevention-and-wellness/family-health-history/index.html>

Does It Run In the Family? toolkit

[Does it Run In the Family? toolkit](http://www.geneticalliance.org/sites/default/files/GuideToFHH/GuidetoFHH.pdf)

<http://www.geneticalliance.org/sites/default/files/GuideToFHH/GuidetoFHH.pdf>



Literacy/Education Resources




Ethics and Privacy



Societal Concerns

- Who should have access to personal genetic information, and how will it be used?
- Who owns and controls genetic information?
- How does personal genetic information affect an individual and society's perceptions of that individual?
- What are the larger societal issues raised by new reproductive technologies?
- How will genetic tests be evaluated and regulated for accuracy, reliability and utility?
- How do we prepare healthcare professionals and the public?
- What is considered acceptable diversity?
- Where is the line between medical treatment and enhancement?
- Should testing be performed when no treatment is available?

GINA

**GENETIC INFORMATION
NONDISCRIMINATION ACT**





About | Contact

Genetic Information
What is genetic information and why is it important?

GINA & Health Insurance
What are GINA's health insurance protections?


GINA & Employment
What are GINA's employment protections?

What is GINA?
The Genetic Information Nondiscrimination Act of 2008 (GINA) is a federal law that protects individuals from genetic discrimination in health insurance and employment. Genetic discrimination is the misuse of genetic information. This resource provides an introduction to GINA and its protections in health insurance and employment. It includes answers to common questions and examples to help you learn. Choose from one of the boxes to the left to begin!





 Have questions, comments or suggestions? [Send us a note.](#)
 [Click here](#) for a printer friendly version.
 For healthcare provider resources [click here](#).
 [Click here](#) for the GINA & You Information Sheet

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National Human Genome Research Institute

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Issues in Genetics


Policy, legal and ethical issues in genetic research




Coverage and Reimbursement of Genetic Tests
Information about insurance coverage for genetic testing




Human Subjects Research
Human subject participation for biomedical, clinical and social-behavioral research




Genetic Discrimination
How Americans are protected from discrimination based on their genetics




Regulation of Genetic Tests
How the federal government regulates genetic tests.




Privacy in Genomics
How best to ensure that genomic information remains private




Informed Consent
The rights of participants when consenting to research projects



Intellectual Property and Genomics
Can a gene be patented?



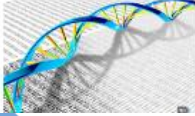
Genetics and Public Policy Fellowship
A fellowship for genetics professionals interested in public policy



Genome Statute and Legislation Database
A database of state statutes and bills from 2007-2016 U.S. state legislative sessions

Highlights

FDA requests comments on draft guidance for Precision Medicine Initiative



The U.S. Food and Drug Administration (FDA) has announced two draft guidances to support President Obama's Precision Medicine Initiative. The guidances will help provide oversight for tests based on next generation sequencing.

See Also

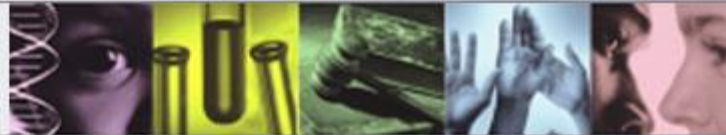
- Policy and Program Analysis Branch
Staff Contact Information
- Ethical, Legal and Social Implications Research Program
NHGRI's Extramural Research Program
- GenomeTV



Informing the Public



CENTER FOR
GENETICS AND
SOCIETY



Precision Medicine

“...a bold new research effort to revolutionize how we improve health and treat disease.”



Precision Medicine Initiative

Mission statement:

To enable a new era of medicine through research, technology, and policies that empower patients, researchers, and providers to work together toward development of individualized care.



[PMI announcement](https://www.whitehouse.gov/precision-medicine) <https://www.whitehouse.gov/precision-medicine>

Precision Medicine is...

- Precision medicine is an emerging approach for disease treatment and prevention that takes into account individual variability in genes, environment, and lifestyle for each person.
- Instead of what treatment is right for this disease it is what treatment is right for this patient.



Precision Medicine Initiative

- **Near Term goals:**
 - Clinical trials focusing on pediatric cancers and drug therapies for adults
 - Use of combination therapies
 - Overcoming drug resistance
- **Long Term Goals:**
 - Create research cohort of 1 million volunteers
 - New model of medicine
 - engage participants
 - responsible data sharing
 - privacy protection
 - Advance pharmacogenomics
 - Identify new targets for treatment and prevention
 - Test if mobile devices encourages healthy behaviors
 - Lay scientific foundation for many diseases

[All of Us infographic:](https://www.nih.gov/sites/default/files/research-training/initiatives/pmi/allofus-inforgraphic-20161117.pdf)

<https://www.nih.gov/sites/default/files/research-training/initiatives/pmi/allofus-inforgraphic-20161117.pdf>

All of Us Research Program

Two ways to participate:

1. Through the participant website
2. With participating Healthcare Provider Organizations (HPOs)



All of UsSM Research Program

WHAT IS IT?

Precision medicine is a groundbreaking approach to disease prevention and treatment based on people's individual differences in environment, genes and lifestyle.

The *All of Us* Research Program will lay the foundation for using this approach in **clinical practice**.

WHAT ARE THE GOALS?

Engage a group of **1 million or more U.S. research participants** who will share biological samples, genetic data and diet/lifestyle information, all linked to their electronic health records. This data will allow researchers to develop more precise treatments for **many diseases and conditions**.

Pioneer a new model of research that emphasizes **engaged research participants, responsible data sharing and privacy protection**.

Research based on the cohort data will:

- Lay **scientific foundation** for precision medicine
- Help identify new ways to **treat and prevent disease**
- Test whether **mobile devices**, such as phones and tablets, can encourage healthy behaviors
- Help develop the **right drug** for the **right person** at the **right dose**

WHY NOW?

The **time is right** because:

- We have a greater understanding of human genes
- People are more engaged in healthcare and research
- We have the tools to track health information and use large databases
- Research technologies have improved

Follow the Program's progress and be one of the first to join this landmark effort.

www.nih.gov/AllOfUs-Research-Program

NIH and Precision Medicine Initiative



The screenshot shows the NIH All of Us Research Program website. At the top is the NIH logo and navigation links. The main header reads "ALL OF USSM RESEARCH PROGRAM". The left sidebar contains links for "All of Us Research Program", "Scale and Scope", "Participation", "Program Components", "Funding", "FAQ", "Advisory Groups", "Events", "Announcements", "In the News", and "Multimedia". Below this is an "All of Us Infographic" section. The main content area features a large graphic with the text "All of Us THE FUTURE OF HEALTH BEGINS WITH YOU" and "The Precision Medicine Initiative". Below this is a section titled "About the Precision Medicine Initiative®" with a paragraph explaining the initiative's goals and a link to a report. To the right of the main content is a sidebar with "Email Updates" and "Related Links" including "PMI Working Group Final Report", "NEJM Perspective: A New Initiative on Precision Medicine", "White House Precision Medicine Web Page", "White House Fact Sheet: President Obama's Precision Medicine Initiative", "Precision Medicine Initiative and Cancer Research", and "Precision Medicine Initiative YouTube Channel".

U.S. Department of Health & Human Services

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Turning Discovery Into Health

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ALL OF USSM RESEARCH PROGRAM

All of Us Research Program

- Scale and Scope
- Participation
- Program Components
- Funding
- FAQ
- Advisory Groups
- Events
- Announcements
- In the News
- Multimedia

All of Us Infographic

All of UsSM Research Program

WHAT IS IT?

Precision medicine is a groundbreaking approach to disease prevention and treatment based on people's individual differences in environment, genes and lifestyle. The All of Us Research Program will lay the foundation for using this approach to individual precision.

WHAT ARE THE GOALS?

Engage a group of 1 million or more U.S. research participants who will share biological samples, genetic data, and detailed lifestyle information, all linked to their personal health records. This data will allow researchers to develop more precise treatments for many diseases and conditions.

Power a new model of research that involves engaged research participants, responsible data sharing, and privacy protection.

All of Us The Precision Medicine Initiative[®]

THE FUTURE OF HEALTH BEGINS WITH YOU

PMI Cohort Program announces new name: the All of Us Research Program

We want to hear from you. Tell us what you think the PMI All of Us Program can achieve.

About the Precision Medicine Initiative[®]

Far too many diseases do not have proven preventions or treatments. To make a difference for the millions of Americans who suffer from them, we must gain better insights into the biological, environmental, and behavioral factors that drive these diseases. Precision medicine is an emerging approach for disease treatment and prevention that takes into account individual variability in environment, lifestyle and genes for each person.

On January 20, 2015, President Obama announced the Precision Medicine Initiative[®] (PMI)[®] in his State of the Union address. As part of PMI, the NIH is leading the effort to build a national, large-scale research enterprise with one million or more volunteers to extend precision medicine to all diseases. The All of UsSM Research Program, formerly known as the PMI Cohort Program, will be a participant-engaged, data-driven enterprise supporting research at the intersection of lifestyle, environment, and genetics to produce new knowledge with the goal of developing more effective ways to prolong health and treat disease. To reflect the diversity of the U.S. population, the program will enroll participants from diverse social, racial/ethnic, ancestral, geographic, and economic backgrounds, from all age groups and health statuses. Information from the program will be a broad, powerful resource for researchers working on a variety of

Email Updates

Sign up to receive email updates about the Precision Medicine Initiative.

[Sign up for updates](#)

Related Links

- [PMI Working Group Final Report](#)
- [NEJM Perspective: A New Initiative on Precision Medicine](#)
- [White House Precision Medicine Web Page](#)
- [White House Fact Sheet: President Obama's Precision Medicine Initiative](#)
- [Precision Medicine Initiative and Cancer Research](#)
- [Precision Medicine Initiative YouTube Channel](#)

[All of Us Research Program:](https://www.nih.gov/research-training/allofus-research-program)

<https://www.nih.gov/research-training/allofus-research-program>

MedlinePlus Magazine- Fall 2015



Health Care Tailored to You

PNR Rendezvous

**Adventures in Precision
Medicine: A Major Public
Research Initiative and its
Implications for Healthcare
Consumers and Institutions**
September 21, 2016



Presenter: Malia Fullerton,
Associate Professor of Bioethics and
Humanities at the University of
Washington School of Medicine



Library role

“Preparing the public to make educated personal and family health decisions in a time of rapidly evolving genetic and genomic knowledge will require new partnerships between the education system, health care systems, the government, community advocacy organizations, consumers and the media.”



[“What Does it Mean to be Genomically Literate? National Human Genome Research Institute Meeting Report”](https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4115323/)

<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4115323/>

Show What You Know!

- What initiative refers to strategies for determining what treatment is right for an INDIVIDUAL rather than what treatment is recommended for a DISEASE?
- What is the name of the volunteer research program that is looking to collect data on 1 million volunteers to assist with the Precision Medicine Initiative?
- True or False? GINA (Genetic Information Nondiscrimination Act) protects you from life insurance discrimination.
- What resource would you recommend to consumers who want to learn more about a genetic testing?



Questions?

Carolyn Martin, MLS, AHIP
Consumer Health Coordinator
NN/LM PNR
martinc4@uw.edu

Presentation resources

[https://nnlm.gov/pnr/guides/
training-resources-you-can-use/presentations](https://nnlm.gov/pnr/guides/training-resources-you-can-use/presentations)

